CLAIMS

We claim:

1. A method of diagnosing CHILD syndrome in a patient comprising the steps of:

(a) isolating patient NSDHL polynucleotide; and

(b) detecting a nucleotide difference between patient NSDHL polynucleotide and the wild type NSDHL gene.

2. A method of diagnosing CHILD syndrome comprising the steps of:

(a) isolating body fluid or cells from a patient; and

(b) detecting in the body fluid or cells accumulation of a sterol intermediate or metabolite thereof prior to the step of generation of cholest-8(9)-en-3β-ol in the cholesterol biosynthetic pathway.

3. The method of claim 2 wherein the sterol intermediate detected is selected from the group consisting of 4,4-dimethylcholest-8(9)-en-3β-ol, 4-methylcholest-8(9)-en-3β-ol, 4-methylcholesta-8(9),24-dien-3β-ol and metabolites thereof.

4. A method of diagnosing psoriasis comprising the steps of:

(a) isolating body fluid or cells from a patient; and

(b) detecting in the body fluid or cells accumulation of a sterol intermediate or metabolite thereof prior to the step of generation of cholest-8(9)-en-3β-ol in the cholesterol biosynthetic pathway.

5. The method of claim 4 wherein the sterol intermediate detected is selected from the group consisting of 4,4-dimethylcholest-8(9)-en-3β-ol, 4-methylcholest-8(9)-en-3β-ol, 4-methylcholesta-8(9),24-dien-3β-ol and metabolites thereof.